Opportunity for Cancer Prevention: 1 in 4 Unaffected Women Meet Hereditary Cancer Testing Criteria

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Disclosure: All authors are current or former employees of Myriad Genetics and/or Myriad Women's Health

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BACKGROUND

- Estimates show 5-10% of cancer is hereditary. Such estimates are derived from populations affected with cancer; the prevalence of hereditary cancer syndromes is not well defined in the unaffected population.
- Approximately 1 in 500[1] individuals in the general population have mutations in *BRCA1* or *BRCA2* and 1 in 300[2] have mutations in genes associated with Lynch Syndrome.
- ACOG states OB/GYNs play an important role in identifying hereditary cancer syndromes (HCS) and recommends standardization for each patient to optimize identification.[3,4]
- This study describes the prevalence of common hereditary cancer syndromes among unaffected female patients as measured by meeting National Comprehensive Cancer Network (NCCN) criteria[5] for genetic testing, following use of a standardized digital identification (ID) tool to capture personal or family cancer history.

METHODS

4226 patients were sent personalized link via SMS or e-mail to a validated digital ID tool[6] prior to their annual well visit with an OB/GYN

Patient completed prompts regarding personal and/or family history of cancer related to NCCN criteria for BRCA-related, Lynch, and Polyposis syndromes

- *How would you describe your ethnic background?*
- *Have you ever been diagnosed with cancer?*
- *Is there a history of cancer in your family?*
- *Has anyone else in your family been diagnosed with cancer?*
- *Have you had any colon polyps?*
- *Has a family member had colon polyps?*
- *Has anyone in your family tested positive for a cancer-related gene mutation?*

Responses were collected between February 1, 2018 and September 1, 2018. Data were de-identified and analyzed to determine how many patients met NCCN criteria as stratified by personal cancer history status.

RESULTS

Figure 1: Distribution of Ethnicities for Unaffected Patients Completing Digital ID Tool (N=2199).

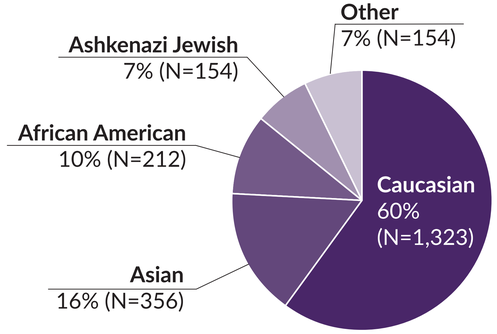
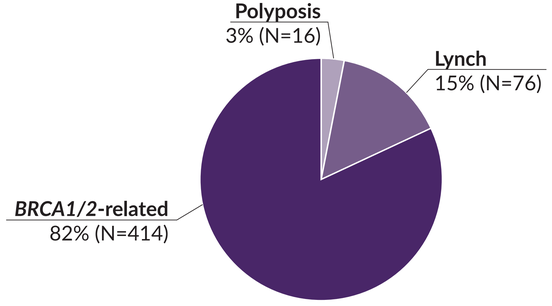


Figure 2: NCCN Testing Criteria Types Met by Unaffected Patients (N=506).



COHORT DEMOGRAPHICS, COMPLETION RATES

- 55% (n=2334) of patients completed the digital ID tool.
- 94% (n=2199) of respondents did not report a personal history of cancer ("unaffected") and had and average age of 42 years.
- The majority of unaffected patients were Caucasian (Figure 1).

CRITERIA INSIGHTS

- Without accounting for known family mutations, 23% (n=506) of unaffected patients met NCCN criteria for hereditary cancer testing across criteria types.
- Breast and gyn-cancer criteria were heavily favored compared to colorectal-related criteria (Figure 2).

CRITERIA NUANCES

- An additional 1.3% met criteria based on known familial mutation to bring the total meets-criteria rate to 24.3%.
- Known family mutations had similar distributions to criteria distributions: 82% had known mutations in *BRCA1* or *BRCA2*, 13% had mutations in genes associated with Lynch Syndrome, and 6% had had *APC* or *MUTYH* mutations.
- 6.5% met criteria for multiple syndromes, all of whom met criteria for both *BRCA1/2*-related and Lynch Syndrome testing, some in part due to known family mutations.

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CONCLUSIONS

- Nearly 1 in 4 unaffected patients presenting for annual OB/GYN visits met NCCN genetic testing criteria for hereditary cancer syndromes, namely *BRCA1/2*-related and Lynch Syndromes.
- Patients in this cohort skewed towards meeting criteria for *BRCA1/2*-related syndromes. This suggests that patients may benefit from education about colorectal cancer, particularly the importance of reporting colorectal-related family history to guide individualized care for cancer prevention including genetic testing. This may also reflect the fact that *BRCA1/2*-related guidelines have expanded more rapidly than Lynch Syndrome guidelines in recent years.
- Standardized interventions to assess family history, such as a digital ID tool, may facilitate identification of patients at risk for hereditary cancer syndromes.

REFERENCES

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